S1 Text. Score statistic

The minor allele frequencies π_1 and π_0 for cases and controls, respectively, are re-parameterized as $\pi_{D_i} = e^{\alpha+\beta D_i}/(1+e^{\alpha+\beta D_i})$ for individual i. Denote $p_i = P_{\epsilon_{D_i}}(R|T, G=0) + 2e^{\alpha+\beta D_i} P_{\epsilon_{D_i}}(R|T, G=1) + e^{2\alpha+2\beta D_i} P_{\epsilon_{D_i}}(R|T, G=2)$ and $q_i = 2e^{\alpha+\beta D_i} P_{\epsilon_{D_i}}(R|T, G=1) + 2e^{2\alpha+2\beta D_i} P_{\epsilon_{D_i}}(R|T, G=2)$. The likelihood function (2) becomes $L_{\mathrm{CC}}(\alpha, \beta, \epsilon_1, \epsilon_0) = \prod_{i=1}^n p_i/(1+e^{\alpha+\beta D_i})^2$ and the score functions with respect to β and α are $S_\beta = \sum_{i=1}^n D_i \left\{q_i/p_i - 2e^{\alpha+\beta D_i}/(1+e^{\alpha+\beta D_i})\right\}$ and $S_\alpha = \sum_{i=1}^n \left\{q_i/p_i - 2e^{\alpha+\beta D_i}/(1+e^{\alpha+\beta D_i})\right\}$, respectively. Under the null hypothesis that $H_0 : \beta = 0$, the score functions S_β and S_α becomes $\sum_{i=1}^n D_i q_i/p_i - 2n_1 e^\alpha/(1+e^\alpha)$ and $\sum_{i=1}^n q_i/p_i - 2ne^\alpha/(1+e^\alpha)$, respectively. Then, we deduce that $S_\beta = S_\beta - (n_1/n)S_\alpha + (n_1/n)S_\alpha = \sum_{i=1}^n (D_i - n_1/n) q_i/p_i + (n_1/n)S_\alpha$. When the nuisance parameters α, ϵ_1, and ϵ_0 are substituted by their restricted MLEs, q_i/p_i is exactly $\widetilde{G}_i$ as defined below equation (3) and $S_\alpha = 0$ by the definition of restricted MLEs. Therefore, we have shown that the score statistic takes the form in equation (3).